

MODELLO SCHEDA RELAZIONE DOTTORANDI
(da redigere in lingua inglese)

Al collegio docenti del Dottorato in Medicina Molecolare

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Ciclo XXXVIII Tutor Prof.ssa Betti Giusti

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During this first year of research activity at the Doctorate in Molecular Medicine, I have been involved in some projects within my working group at the Atherothrombotic Disease Centre of the Department of Experimental and Clinical Medicine of the University of Florence. A study investigates the genetic causes of BAV, this valvular malformation is due to an error in foetus development; in the majority of cases, the defect in question is the result of a hereditary mutation. Different genes have been associated or suspected in BAV pathogenesis and complications. In order to improve knowledge on BAV, NGS sequencing was carried out using Illumina NextSeq500 and MiSeq platform, the libraries were built up using the Agilent SureSelect kit and the corresponding quality control using Bioanalyzer and Qubit on more than 50 subjects. We are now evaluating and classifying the rare genetic variants identified in analysed patients.

Another line of research to which I have contributed concerns the analysis of genetic data obtained from our diagnostic centre concerning familial aortopathies, Marfan disease and related with the aim of identifying further genotype-phenotype correlations. A database was created on the SPSS statistical platform in which rare (MAF less than 1%) missense, splicing and synonymous variants of our custom panel of 102 genes were included. I also contributed to the data analysis of a whole exome of 6 patients with vaccine-induced immune thrombotic thrombocytopenia (VITT). In particular, an enrichment analysis was carried out that did not reveal any pathways or genes significantly associated with VITT, while at the level of the individual variants selected in genes involved in the pathways of haemostasis, fibrinolysis and platelet activation and aggregation, it was observed that in the six patients there are between 2 and 9 variants of uncertain clinical significance (VUS) according to ACMG nomenclature. These data has been submitted for publication.

• Abstracts

- Tommaso Capezzuoli, Rosina de Caro, Elena Sticchi, Marco Giannini, Rebecca Orsi, **Lapo Squillantini**, Samuele Suraci, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Vaccine Induced Thrombotic Thrombocytopenia: Evaluation Of Genetic Susceptibility Throught Whole Exome Sequencing. University of Florence; Dpt. Experimental and Clinical Medicine, Florence. Congresso Nazionale Siset, 2/5 Novembre 2022 Perugia
- **L. Squillantini**, E. Sticchi, T. Capezzuoli, M. Giannini, G. Cassioli, G. Barbieri, S. Suraci, A. M. Gori, M. Berteotti, A. A. Rogolino, R. De Caro, A. Kura, R. Marcucci, B. Giusti - Individual

Vulnerability to SARS-COV-2 infection: genetic profiles and the role of the host's virus entry machine - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023

- G. Barbieri, A.M. Gori, F. Cesari, E. Sticchi, A. Rogolino, R. Orsi, T. Capezzuoli, G. Cassioli, M. Giannini, **L. Squillantini**, A. Bertelli, R. De Caterina, B. Giusti, R. Marcucci - Edoxaban Effects On Platelet Function: In Vitro And Ex Vivo Studies - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023
- T. Capezzuoli, R. De Caro, E. Sticchi, M. Giannini, R. Orsi, **L. Squillantini**, G. Cassioli, G. Barbieri, S. Suraci, A.M. Gori, R. Marcucci, B. Giusti – Vaccine INduced Thrombotic Thrombocytopenia (VITT): Evaluation of Genetic Susceptibility through WES - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023
- M. Giannini, E. Sticchi, T. Capezzuoli, R. De Caro, **L. Squillantini**, R. Orsi, S. Suraci, G. Cassioli, S. Neroni, G. Barbieri, A. A. Rogolino, M. Berteotti, A. M. Gori, R. Marcucci, B. Giusti - Identification of rare coagulation variants and screening of polymorphisms (F2 rs1799963, F5 rs6025) in COVID-19 patients - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023
- G. Cassioli, A. Kura, A. Sodero, E. Sticchi, A. Magi, S. Suraci, **L. Squillantini**, A. Consoli, A. Rosi, S. Nappini, L. Renieri, N. Limbucci, B. Piccardi, F. Arba, C. Sarti, D. Inzitari, S. Mangiafico, R. Marcucci, A. M. Gori, B. Giusti - Gene Expression Profiles According To Etiology In Acute Ischemic Stroke Patients - Corso Nazionale Siset 2023 Foggia 26 - 28 ottobre 2023

• Partecipazioni a congressi e corsi

- 2nd Meeting Translational research on stroke (with Open Discussions on NIMBLE Results) – Firenze, 25 novembre 2022
- Il ruolo degli anticoagulanti iniettivi nel TEV: confronto con gli esperti – Firenze, 2 dicembre 2022
- Congresso Regionale SISA sezione toscano-emiliana – Firenze, 10 dicembre 2022
- Giornata della ricerca Dmsc dal titolo “Il Dmsc presenta i giovani” – Firenze, 22 dicembre 2022
- Dottorato in Medicina Molecolare, PhD Day – Firenze, 17 gennaio 2023
- Il Dmsc dalla parte delle donne, Giornata Internazionale della Donna – Firenze, 8 marzo
- Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023 – Presentazione poster dal titolo “Individual Vulnerability to SARS-COV-2 infection: genetic profiles and the role of the host's virus entry machine”

• Pubblicazioni scientifiche autori, titolo della pubblicazione, nome e numero della rivista, anno di pubblicazione

- Submitted 04/10/2023 - Betti Giusti, Elena Sticchi, Tommaso Capezzuoli, Rebecca Orsi, **Lapo Squillantini**, Marco Giannini, Samuele Suraci, Angela Rogolino, Francesca Cesari, Martina Berteotti, Anna Maria Gori, Elena Lotti, Rossella Marcucci - Whole exome

sequencing in Vaccine-induced thrombotic thrombocytopenia (VITT). Thrombosis and Haemostasis